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AUTHOR Brown, Denice P.
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ABSTRACT

Eight children (ages 13 days to 5 years) with a diagnosis of Cornelia de Lange syndrome received audiologic evaluation consisting of immittance audiometry and auditory brainstem response audiometry to air and bone conducted "click" stimuli, as behavioral testing was unreliable due to patient age and/or developmental delay. Developmental assessments were also carried out on five of the children. Results indicated that all eight children had hearing sensitivity loss ranging from mild to severe and of varied site-of-lesion. Four children with mild conductive hearing loss had a history of chronic otitis media and were subsequently managed with pressure equalization tube placement and/or medication. Three with moderate to severe mixed or sensori-neural hearing loss required hearing aid fitting and special educational placement. The remaining child expired shortly after birth. Results of the developmental assessments found that four of the five children were characterized by severe to profound mental and motor delays. Data showed no relationship between the degree of developmental delay and the presence or absence of a significant hearing sensitivity loss. Case studies of two of the children are presented. (Seven references and 10 tables/figures) (JDD)

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ABR AUDIOMETRY IN CORNELIA DE LANGE SYNDROME

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of
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DENICE P. BROWN, PH.D.
Assistant Professor
University of Houston
Program in Communication Disorders
School of Communication
Houston, Texass 77204-6611
(713) 749-2741

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INTRODUCTION

Cornelia de Lange Syndrome is characterized by growth and mental retardation, failure to thrive, microcephaly, thin lips turned down at angles of the mouth, long philtrum, high-arched palate, low set ears, and hirsutism. Individuals with this syndrome may also exhibit a variety of limb and/or skeletal defects (i.e., proximal implantation of thumbs, clinodactyly, syndactyly), visual defects, cleft palate, genital abnormalities (in males), feeding problems, and speech and language disorders. Reported occurrence is 1 in 50,000, and diagnosis is based solely upon clinical assessment as there are currently no biochemical or chromosomal markers associated with the de Lange Syndrome.

Literature discussing the syndrome has focused primarily on physical exam results as well as a variety of laboratory, radiographic, metabolic, and chromosomal studies. The possibility of hearing sensitivity loss has received little attention, although Hawley, Jackson, and Kurnit (1985) reported hearing loss in 14.3% of 64 individuals surveyed, and Marres, Cremers, and Jongbloet (1989) reported some degree of sensitivity loss in seven individuals evaluated with pure tones in sound field or under earphones. The lack of documented audiologic data may be related to the inability of prior investigators to reliably employ traditional audiologic techniques due to the significant developmental delay and physical handicaps manifest by the majority of these individuals. The advent of Auditory Brainstem Response Audiometry (ABR), however, has provided a means of objectively evaluating hearing status in this unique disorder.

METHOD

Within the past six years, eight children (13 days to 5 years of age) with a diagnosis of Cornelia de Lange syndrome received audiologic evaluation. Initial assessments typically consisted of immittance audiometry and auditory brainstem response audiometry (ABR) to air and bone conducted "click" stimuli since behavioral testing was unreliable due to patient age and/or developmental delay. Developmental assessments were also carried out on five of the eight children by a child development specialist.

RESULTS

Hearing sensitivity loss ranging from mild to severe degree and of varied site-of-lesion was noted in all eight children on initial assessment. Table I shows the degree of hearing sensitivity loss and corresponding site-of-lesion for the eight individuals. The four children with mild conductive hearing loss had a history of chronic otitis media and were subsequently managed with pressure equilization (PE) tube placement and/or medication. Three with moderate to severe mixed or sensori-neural hearing loss required hearing aid fitting and special educational placement. The remaining child with an apparently severe hearing sensitivity loss expired shortly after birth.

Developmental assessments were carried out on five of the eight children. As illustrated in Table II, evaluation results were characterized by severe to profound mental and motor delays in all but one child. These data, however, show no relationship between the degree of developmental delay and the presence or absence of a

significant hearing sensitivity loss as determined by ABR. Of the three children not receiving developmental evaluations, one expired and the remaining two are reported to be severely delayed.

ILLUSTRATIVE CASES

CASE STUDY 1 - E.J.

EJ was a low birthweight, full-term, microcephalic infant who spent 2 months in the Neonatal Intensive Care Unit (NICU) for ventilatory support. He was born with a cleft palate, midface hypoplasia, and an underdeveloped mandible. His right arm has a rudimentary hand with two digits; his left arm has a single digit hand. He additionally has a mild form of aortic stenosis. Medical history is significant for chronic otitis media and mutiple PE tube placements.

Developmental assessments were carried out at 22 months, 2 years-4 months, 3 years, and 3 years-6 months of age. All skills developed slowly and plateaued by the age of 3 years. Mental age is in the 12-15 month range, receptive language is in the 2-19 month range, and gross motor skills are in the 7-15 month range.

As shown in Figure 1, ABR assessment to click stimuli at 20 months of age yielded poorly formed air conduction responses of prolonged absolute latency as low as 55 dB nHL and 60 dB nHL for the right and left ears, respectively. An unmasked bone conduction response (non-specific to either ear) was present down to 10 dB nHL. Acoustic immittance measures were consistent with middle ear disorder. Findings were characterized by Type B tympanograms, low static compliance measures, and absent acoustic reflexes. EJ was referred

for otologic management and was subsequently scheduled for bilateral myringotomy and PE tube placement.

Follow-up audiologic assessment was carried out at 23 months of age while PE tubes were in place. ABR air conduction thresholds were elevated in comparison to the previous test. Thresholds were 85 dB nHL for the right ear and 65 dB nHL for the left ear (Figure 2). Ear specific bone conduction waveforms were present down to 35 dB nHL and 5 dB nHL, with right and left mastoid placement, respectively. The intensity-latency function illustrated in Figure 3 is suggestive of a moderate to moderately-severe, primarily conductive hearing sensitivity loss, bilaterally. In view of the unexpected elevation in ABR air conduction thresholds, EJ was referred for otologic re-examination. He was also placed in a parent-infant training program for multiple handicapped children.

EJ did not return for additional audiologic testing until 3 years of age. Patent PE tubes were in place at the time of evaluation. Attempts to carry out behavioral audiometry were unsuccessful due to his severe developmental delay and physical limitations. Air and bone conduction ABR tracings are shown in Figure 4. As these waveforms illustrate, replicable air conduction responses were present as low as 65 dB nHL, bilaterally, and apparently ear specific bone conduction thresholds (contralateral ear masked) were 40 dB nHL and 45 dB nHL for the right and left ears, respectively. The intensity-latency function represented in Figure 5 suggested a bilateral mixed hearing sensitivity loss, and indicated a deterioration in cochlear sensitivity when findings were compared to those acquired previously.

EJ received otologic clearance for hearing aid use and was fitted with appropriate amplification. He is receiving services from an Infant-Parent Training Center for multiple handicapped children. His middle ear status is closly monitored by an otolaryngologist due to recurrent otitis media.

CASE STUDY 2 - J.F.

JF was a full term, low birthweight infant with microcephaly and facies characteristic of Cornelia De Lange Syndrome. There are no obvious limb or skeletal defects.

Developmental assessments were carried out at 2 1/2 months, 6 1/2 months, and 2 years-6 months of age. Initially, developmental skills were superior to those of most Cornelia de Lange infants. By 17 1/2 months, however, a slowing of development was noted. By 2 1/2 years of age, severe mental delay was evident, although gross motor skills were only mildly delayed.

The initial ABR assessment was carried out at 2 1/2 months of age and waveforms are documented in Figure 6. A low amplitude air conduction ABR was observed down to 60 dB nHL and 70 dB nHL for the right and left ears, respectively. In addition, an unmasked bone conduction response was present at 30 dB nHL, suggesting a significant conductive component in at least one ear. Further bone conduction testing and immittance audiometry could not be carried out due to an increase in the infant's activity level. The referring physician did not wish to administer additional sedation at that time.

Behavioral Observation Audiometry at 6 months of age yielded quieting and searching responses to speech stimuli at 80-90 dB HL. ABR Audiometry to air conducted click stimuli only generated responses at 95 dB nHL, bilaterally, and an apparently ear-specific bone conduction response for the right ear was obtained at 50 dB nHL (Figure 7). A response as low as 30 dB nHL was present with left mastoid placement, although it was not possible to ascertain if the response was ear specific due to the lack of a component wave I on the acquired tracings. Furthermore, it was not possible to effectively mask the right ear due to the severity of the loss by air conduction. Attempts to carry out immittance audiometry were once again unsuccessful due to JF's active behavior. Otologic examination, however, failed to demonstrate active middle ear disorder and the patient was cleared for hearing aid use. When fitted with binaural amplification, behavioral responses to speech were observed at 50-60 dB HL.

Audiologic testing at 11 months of age was carried out to monitor hearing status due to the elevation in air conduction thresholds noted on the previous test. As shown in Figure 8, air conduction responses were only observed as low as 90 dB nHL, bilaterally, and an unmasked bone conduction waveform was obtained at 45 dB nHL. In summary, the overall pattern of results suggested a severe hearing sensitivity loss, bilaterally. Bone conduction versus air conduction responses also indicated a substantial conductive component to the loss. JF continues to utilize binaural amplification and is receiving aural habilitation services.

CONCLUSIONS

Results of ABR assessment in children with Cornelia de lange Syndrome indicate a high incidence of hearing sensitivity loss in this population. Permanent congenital hearing loss may be conductive, sensori-neural, or mixed, and there is evidence of a progressive component in some cases. These individuals also tend to experience recurrent bouts of otitis media and concomitant transient conductive hearing loss.

Overall findings thus support the need for complete audiologic assessment of all individuals with a diagnosis of Cornelia de Lange. Close monitoring of hearing status is definitely indicated in view of the high incidence of otits media and the possibility of progessive sensori-neural or conductive sensitivity loss. In addition, ABR audiometry appears to be a viable technique for assessing peripheral hearing sensitivity in individuals with Cornelia De Lange syndrome who are unable to respond to traditional audiologic methods. It is important to recognize the high incidence of hearing loss associated with this syndrome, particularly in view of the fact that the 1990 Position Statement issued by the Joint Committee on Infant Hearing recommends the screening of infants with syndromes known to be associated with hearing loss.

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TABLE/FIGURE LEDGENDS

TABLE I. Degree of hearing sensitivity loss and corresponding site-of-lesion in 8 individuals with Cornelia de Lange syndrome.

TABLE II. Results of developmental assessments in 8 individuals with Cornelia de Lange syndrome.

FIGURE 1. ABR waveforms obtained from EJ at 20 months of age in response to air and bone conducted click stimuli.

FIGURE 2. ABR air and bone conduction waveforms obtained from EJ at 23 months of age following PE tube placement. Note the elevation in air conduction thresholds in comparison to the previous test at 20 months of age.

FIGURE 3. ABR intensity-latency function for EJ at 23 months of age.

FIGURE 4. ABR waveforms obtained from EJ at 3 years of age. Note the elevation in bone conduction thresholds since prior testing at 20 and 23 months.

FIGURE 5. ABR intensity-latency function for EJ at 3 years of age.

FIGURE 6. ABR waveforms obtained from JF at 2 1/2 months of age in response to air and bone conducted click stimuli.

FIGURE 7. ABR waveforms obtained from JF at 6 months of age in response to air and bone conducted click stimuli. Bone conduction thresholds are unmasked since it was not possible to effectively mask the nontest ear due to the significant elevation of ABR air conduction thresholds.

FIGURE 8. ABR waveforms obtained from JF at 11 months of age in response to air and bone conducted click stimuli.

	CONDUCTIVE	MIXED	SENSORI-NEURAL
MILD	*4		
MODERATE		1	1
SEVERE		1	1

*Confirmed otitis media with effusion at time of test

TABLE I.

SUBJECT	MENTAL DELAY	MOTOR DELAY	DEGREE & TYPE HEARING LOSS
J.F.	severe	mild	severe mixed
E.J.	severe	severe	moderate mixed
R.R.	severe	severe	moderate sensori-neural
H.M.	severe	severe	mild conductive (OM)
M.B.	mild	mild	mild conductive (OM)

TABLE II.

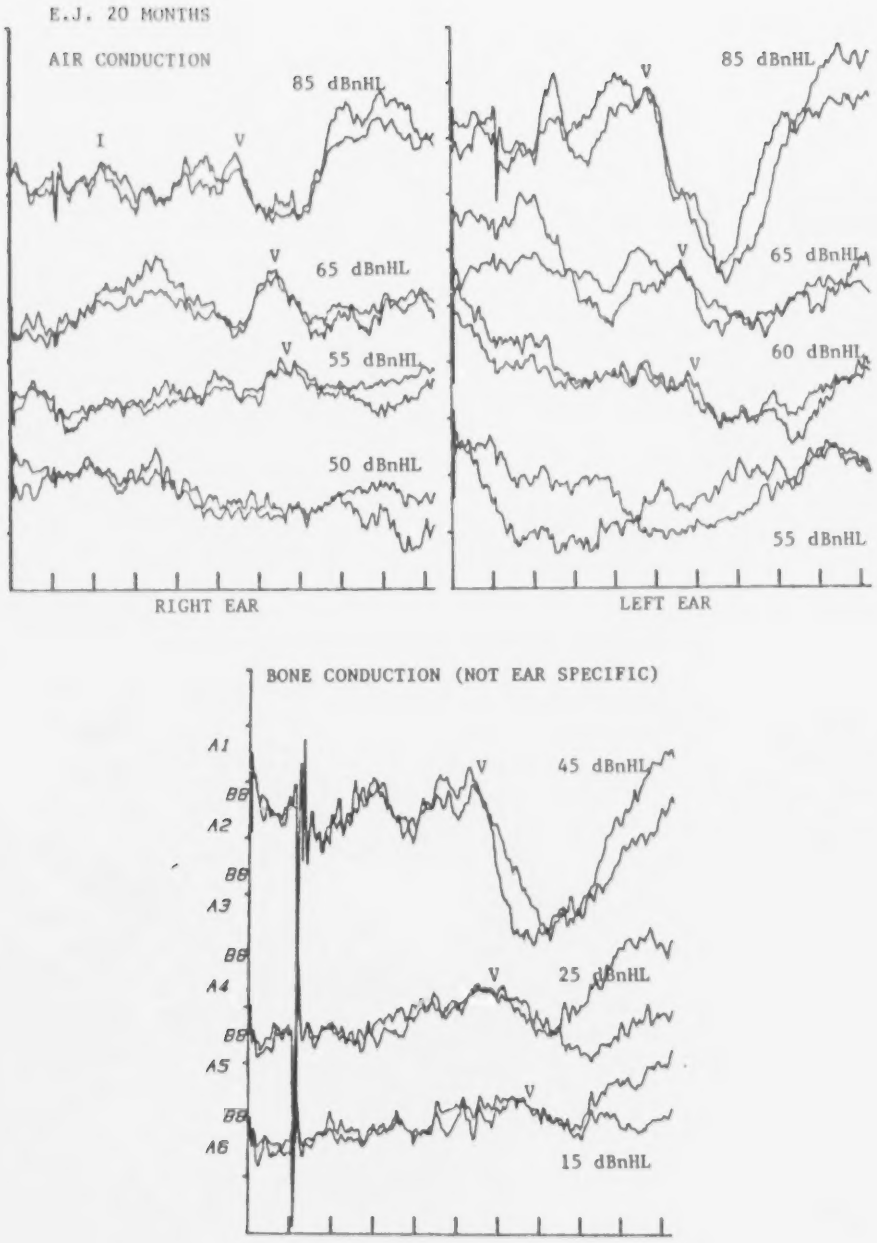
E.J. 20 MONTHS
AIR CONDUCTION
85 dBnHL
I
V
65 dBnHL
55 dBnHL
50 dBnHL
RIGHT EAR
85 dBnHL
65 dBnHL
60 dBnHL
55 dBnHL
LEFT EAR
BONE CONDUCTION (NOT EAR SPECIFIC)
A1
BB
A2
BB
A3
BB
A4
BB
A5
BB
A6
45 dBnHL
25 dBnHL
15 dBnHL

FIGURE 1.

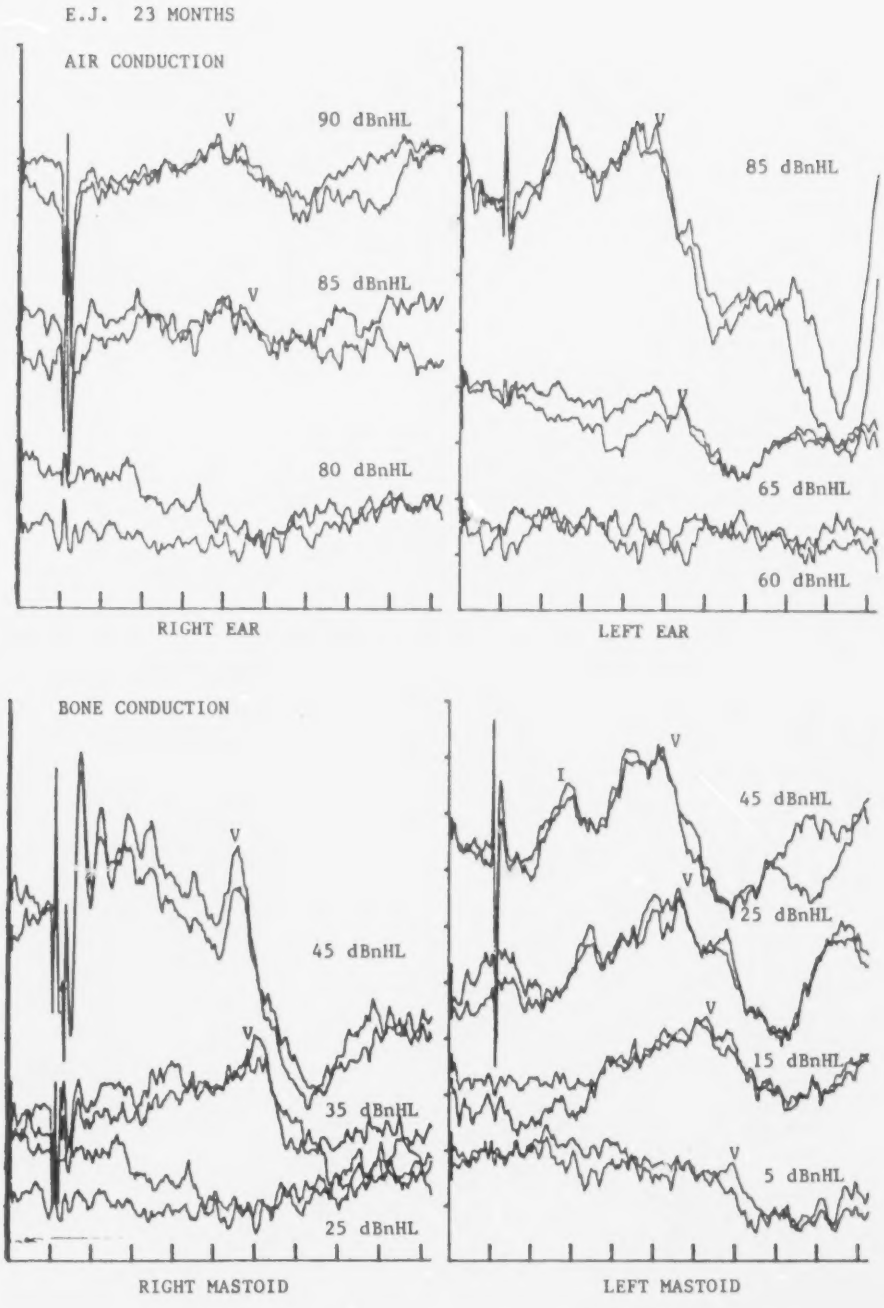
E.J. 23 MONTHS
AIR CONDUCTION
V
90 dBnHL
85 dBnHL
80 dBnHL
85 dBnHL
65 dBnHL
60 dBnHL
RIGHT EAR
LEFT EAR
BONE CONDUCTION
I
45 dBnHL
35 dBnHL
25 dBnHL
25 dBnHL
15 dBnHL
5 dBnHL
RIGHT MASTOID
LEFT MASTOID

FIGURE 2.

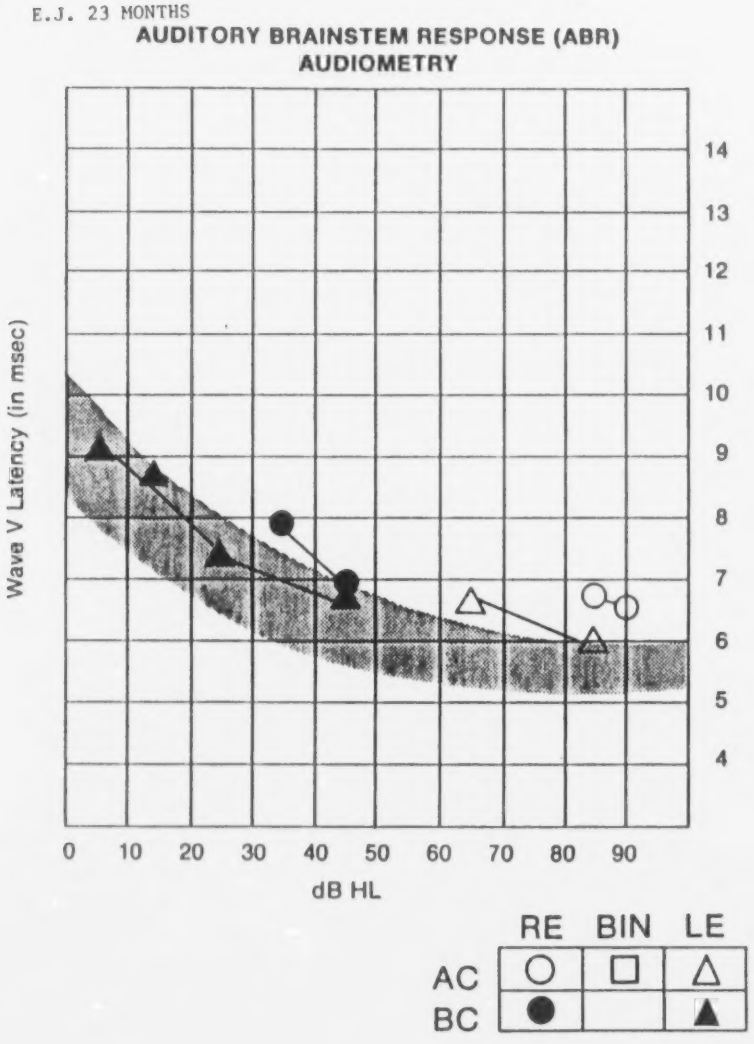
E.J. 23 MONTHS
AUDITORY BRAINSTEM RESPONSE (ABR)
AUDIOMETRY
Wave V Latency (in msec)
14
13
12
11
10
9
8
7
6
5
4
0 10 20 30 40 50 60 70 80 90
dB HL
RE BIN LE
AC
BC

FIGURE 3.

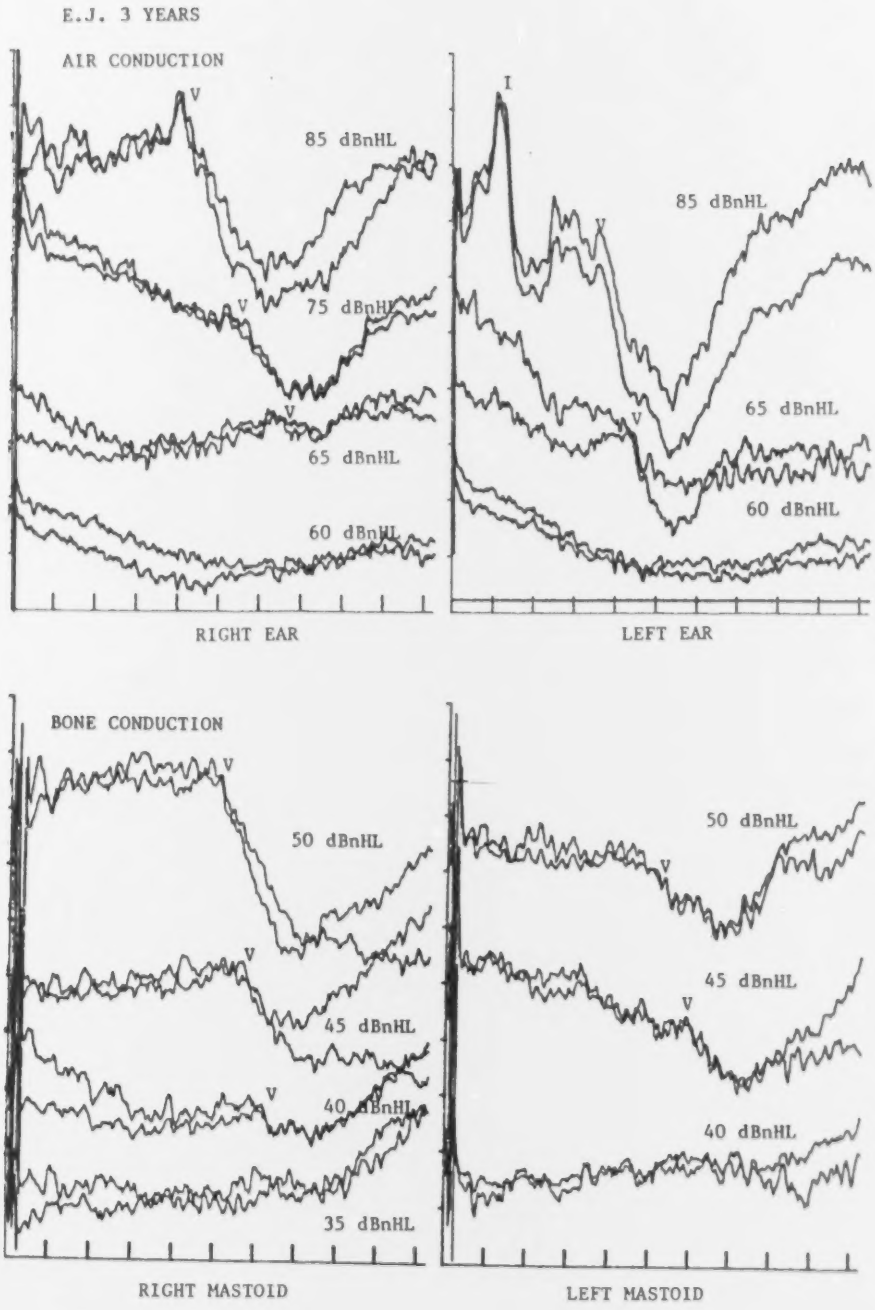
E.J. 3 YEARS
AIR CONDUCTION
V
85 dBnHL
V
75 dBnHL
V
65 dBnHL
60 dBnHL
RIGHT EAR
I
85 dBnHL
V
V
65 dBnHL
60 dBnHL
LEFT EAR
BONE CONDUCTION
V
50 dBnHL
V
45 dBnHL
V
40 dBnHL
35 dBnHL
RIGHT MASTOID
50 dBnHL
V
45 dBnHL
V
40 dBnHL
LEFT MASTOID

FIGURE 4.

E.J. 3 YEARS

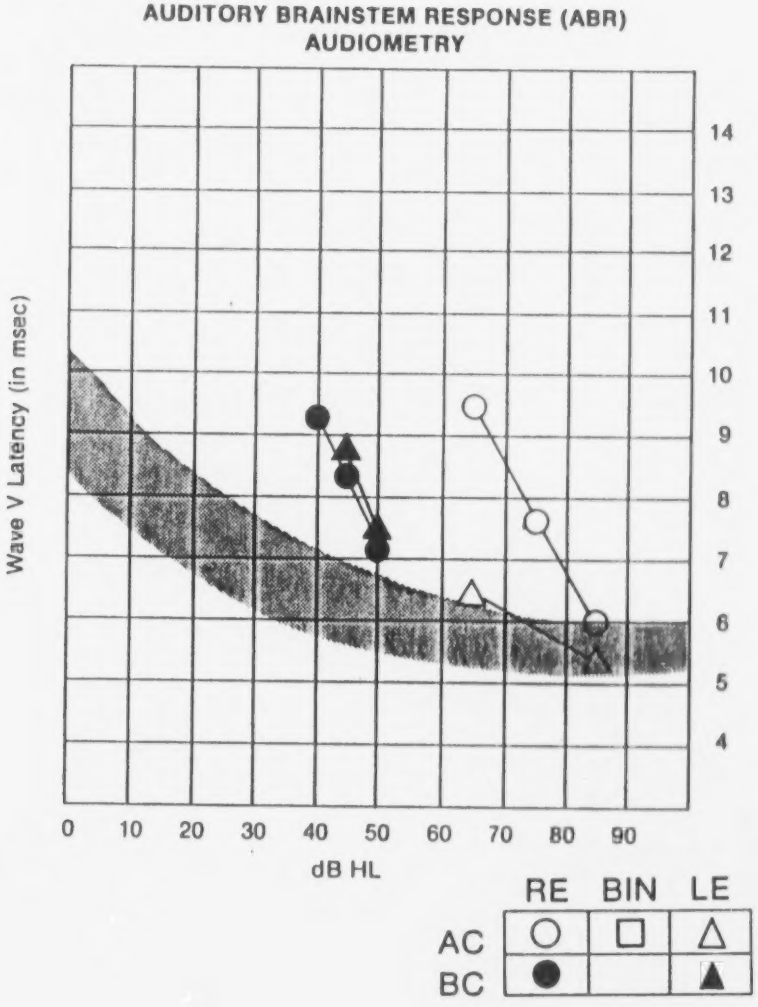

FIGURE 5.

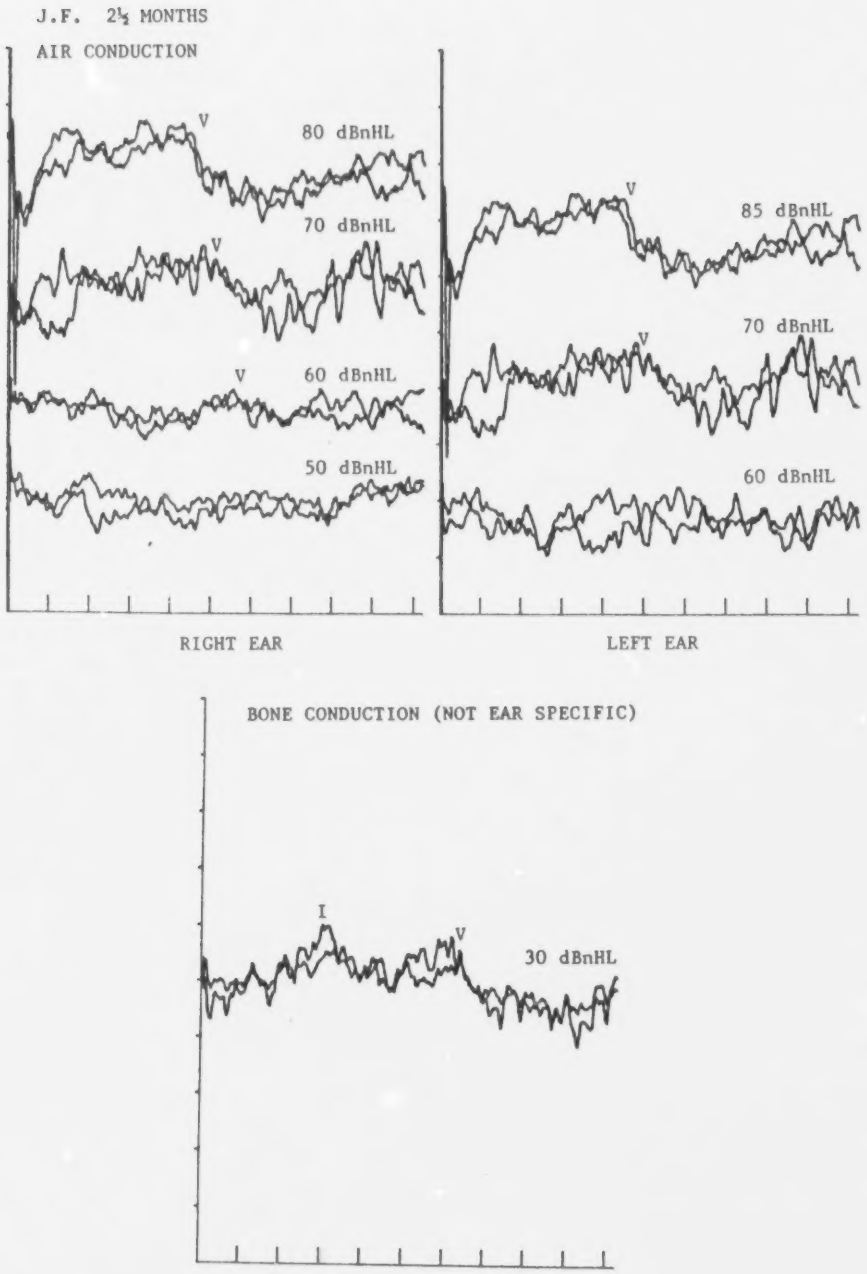
J.F. 2½ MONTHS
AIR CONDUCTION
V
80 dBnHL
70 dBnHL
V
V
60 dBnHL
50 dBnHL
V
85 dBnHL
70 dBnHL
V
60 dBnHL
RIGHT EAR
LEFT EAR
BONE CONDUCTION (NOT EAR SPECIFIC)
I
V
30 dBnHL

FIGURE 6.

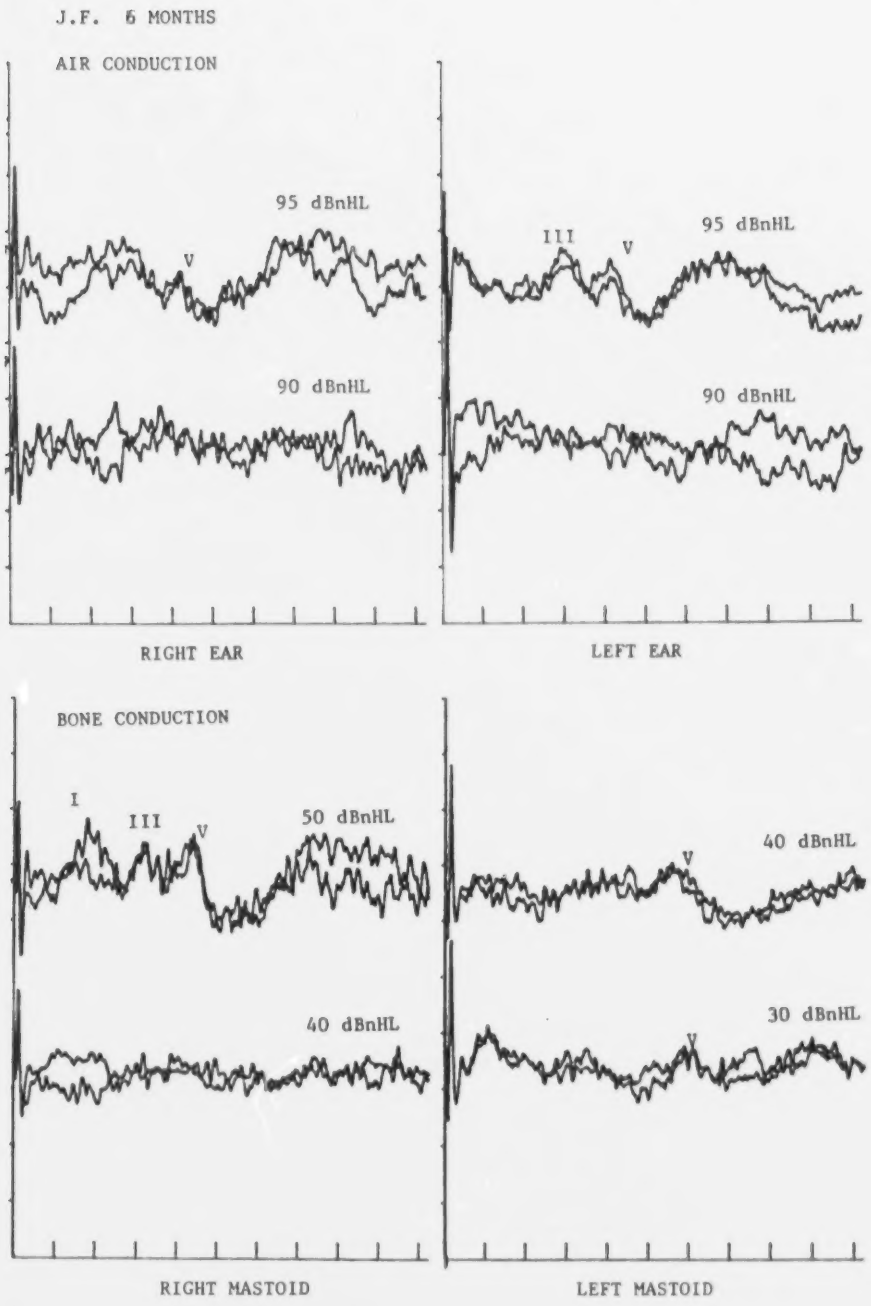
J.F. 6 MONTHS
AIR CONDUCTION
95 dBnHL
V
90 dBnHL
III
V
95 dBnHL
90 dBnHL
RIGHT EAR
LEFT EAR
BONE CONDUCTION
I
III
V
50 dBnHL
40 dBnHL
V
40 dBnHL
30 dBnHL
V
RIGHT MASTOID
LEFT MASTOID

FIGURE 7.

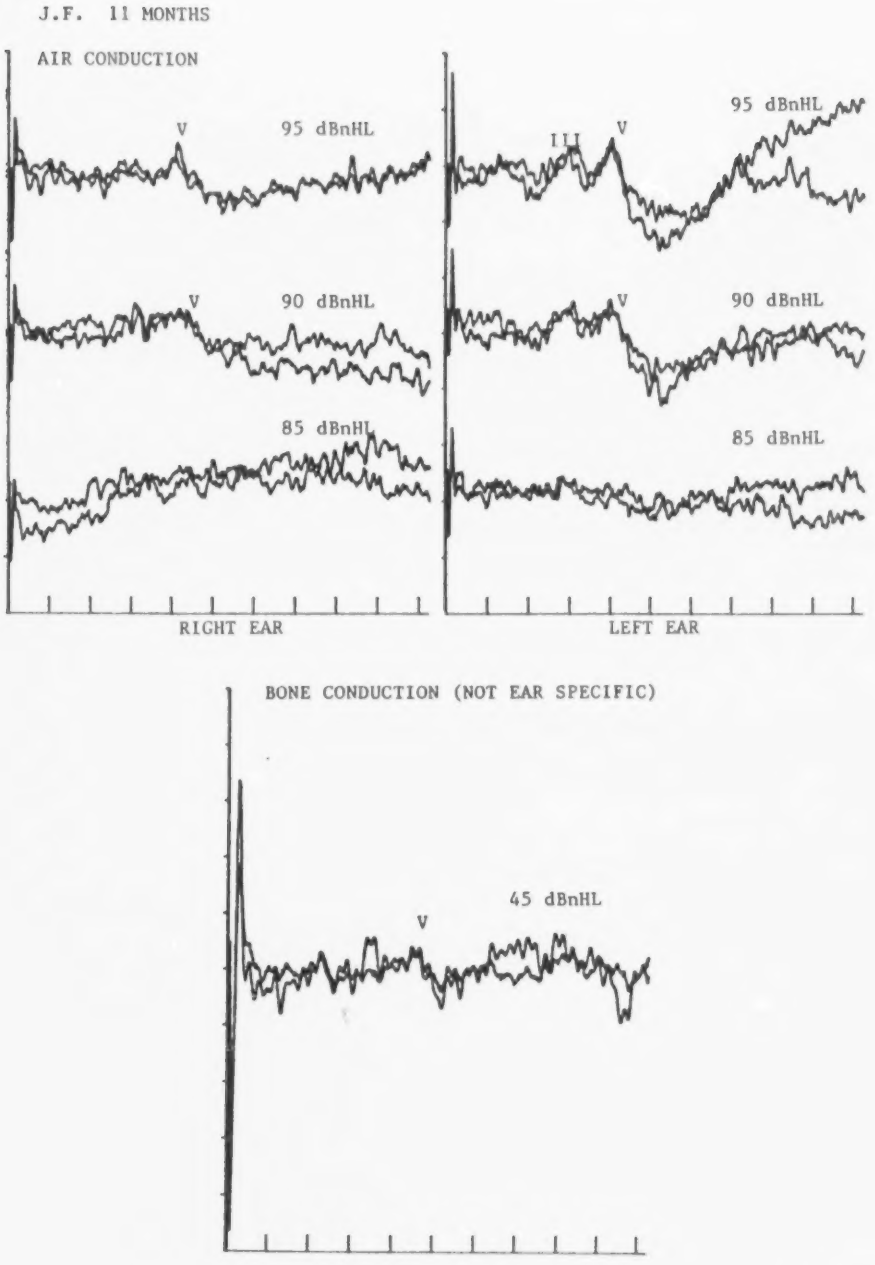
J.F. 11 MONTHS
AIR CONDUCTION
V
95 dBnHL
V
90 dBnHL
85 dBnHL
RIGHT EAR
III
V
95 dBnHL
V
90 dBnHL
85 dBnHL
LEFT EAR
BONE CONDUCTION (NOT EAR SPECIFIC)
V
45 dBnHL

FIGURE 8